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Clinical Records

The Scheibe cochlea deformity with macrocephaly: a case for single channel implantation

J. Ray, R. F. Gray, Z. H. Vanat, J. A. Begg

Abstract
An 11-year-old congenitally deaf child with bilateral primitive common cavity (Scheibe type) cochleosaccular dysplasia and benign familial macrocephaly was implanted with an extracochlear single channel device with an ear level speech processor. This paper describes the assessment, findings, dilemmas in decision making, surgical procedure and the favourable outcome after implanting. The relevant literature has been reviewed and our case is presented for the unusual combination of features.

Key words: Cochlear implant; Cochlea; Abnormalities; Deafness, congenital

Introduction

Congenital anomalies of cochlear morphology are well-known causes of profound sensorineural deafness. An 11-year-old boy from a farming family with Scheibe type cochleo-saccular dysplasia was assessed and found to be profoundly deaf in both ears beyond the reach of hearing aids but not suitable for a multichannel cochlear implant because of (1) lack of tonotopic representation and (2) middle-ear sepsis with drum perforation and discharge. He was tried with a vibrotactile device and later implanted with an extracochlear single channel device to augment his lip-reading skills and provide awareness of environmental sounds.

The unusual association of sensorineural deafness due to cochlear dysmorphology with hypotonia and autosomal dominant benign familial macrocephaly aroused our interest.

Case report

The child was first suspected to be deaf by the childminder at one year old when there was no response to a door slamming shut. After failing three health visitor distraction tests he was referred to the local ENT department where bilateral post-aural hearing aids were fitted at the age of 18 months.

Reassessment at two years showed very little language development (only single indistinct words). Tympanograms were flat and responses on electrocochleography were equivocal at 95 dBA. At this stage grommets were fitted. These were later replaced by T-tubes. Repeated ear infections added to the problems and they were removed after four years leaving small perforations which were still discharging and troublesome when assessment for cochlear implant was in progress. These eventually closed over.

Associated features

The patient was the elder child of parents with normal hearing and without any family history of deafness. He was a normal term birth but was thought to have infantile hypotonia. It was also noted that the head circumference was 1 standard deviation (SD) greater than the mean for his age and this continued to increase steadily until it was

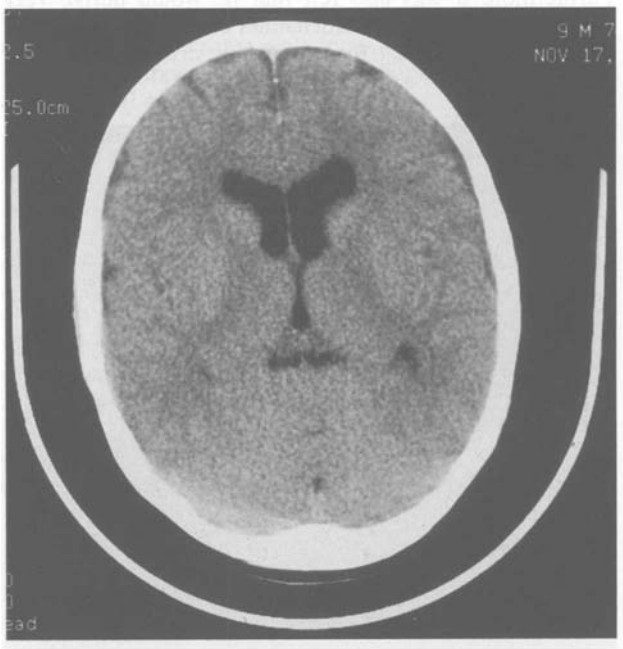

Fig. 1
CT scan of the patient's brain showing slight dilatation of lateral ventricles.

From the East of England Cochlear Implant Programme, Addenbrooke's Hospital, Cambridge, UK.
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greater than 4 SD of the mean at four years of age. A computed tomography (CT) scan showed enlarged lateral ventricles (Figure 1). A review of the family tree and CT scanning of the parents (when similar findings were seen in the father) by the Medical Genetics Department revealed the existence of a rare disorder – benign familial macrocephaly. This was inherited as an autosomal dominant disorder from the father's side. The head growth stopped at 4 SD from the mean head circumference (findings in his father were the same). Interestingly, from the mother's side the child had inherited an idiopathic ptosis on the right eye which had been present through at least three generations. Other somatic abnormalities seen in the child were increased distance between the medial canthus with downslanting palpebral fissures, an upturned nose, broadened forehead and a flat midface. His motor skills were rather delayed and he learned to walk only at the age of three years. Thereafter he made steady progress and by seven years he was riding a bicycle without stabilizers.

Initial referral

The patient was referred to our assessment clinic at the age of nine years for consideration of cochlear implantation. Ultra-high resolution CT scanning showed 'a widened and dysplastic basal turn of the cochlea without any round window or promontory which in turn was separated from the internal auditory meatus (IAM) by a thin bony septum (Schiebe type cochleosaccular dysplasia). Semicircular canals also appeared to be hypoplastic. The carotid canals were prominent bilaterally and only just separated from the middle ear cavity by a thin bony septum' (Figure 2). Audiological results were as seen in Table I.

An intracochlear implant was not thought to be advisable because of the risks of a CSF leak (Phelps *et al.*, 1993) compounded by chronic middle ear sepsis. Furthermore it was also felt that he would derive very limited benefit from a multichannel cochlear implant as it was not expected that those cochlear structures that had developed would be organized in the usual tonotopic way. The child was, therefore, fitted with a TRILL vibrotactile aid.

TABLE I
UNAIDED PURE TONE AUDIOGRAM

Frequency (Hz)	250	500	1K	2K	4K
dBHL Left ear	110	95	120	115	NR
dBHL Right ear	90	95	115	115	NR

Reassessment

At one year he was reassessed and found not to be using the TRILL at all as he found the waist-worn box attached to wristbands very cumbersome. Ear infections had settled by then and the perforations had closed over with granular myringitis which could be improved by topical antibiotic cream. Promontory stimulation and auditory brainstem response tests showed a few inconsistent responses. Detailed 0.7 mm section T2-weighted fast spin echo MR scans were done to demonstrate the anatomical integrity of the auditory nerve pathway from the internal auditory meatus (Figure 3). This showed 'bilateral primitive common cavity lesions on both sides but no definable cochlea. However there were several nerves going through the IAMs'.

Further testing was carried out to confirm the functional value of his communication. He was performing well on lip reading screens but was unable to detect any meaning from voice alone.

Device selection

Experience with similar pathologies at other centres in Europe and North America was reviewed and it was decided to recommend a single channel extracochlear device. The parents were counselled. They were sure that a body worn device would be rejected and that the child would only take to an ear level device. The Med El Extracochlear implant with an ear level single channel speech processor (being the only ear level device available then) was chosen. The right ear was selected for implantation.

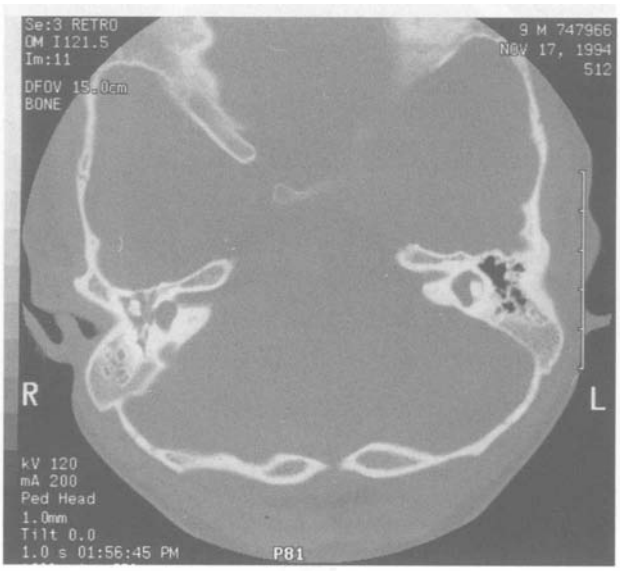

FIG. 2
CT scan (1 mm axial sections) of the patient showing bilateral cochleosaccular dysplasia and internal auditory meatus separated from the middle ear by a thin bony partition.

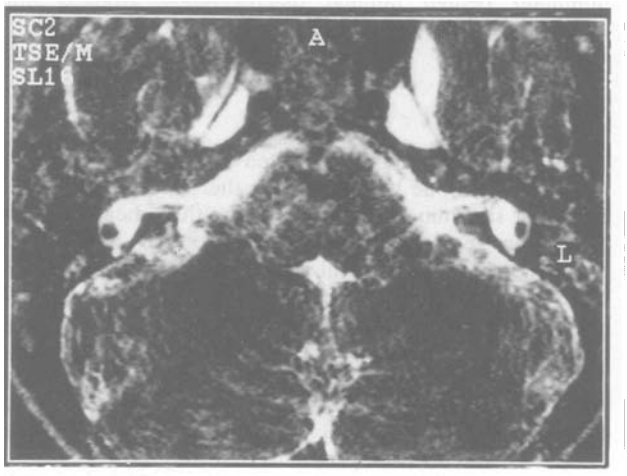

FIG. 3
Axial 0.7 mm T2-weighted MR image of IAMs showing existence of neural structures in both IAMs and leading up to a primitive common cavity.

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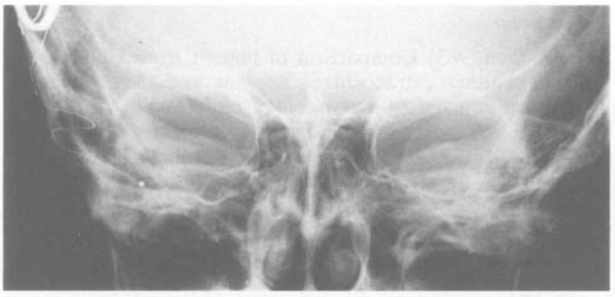

FIG. 4
Post-operative perorbital skull X-ray of the patient showing position of the single channel extracochlear ball electrode.

Surgery

The usual approach for a cochlear implant was used with no complications. A high promontory was visualized but no identifiable round window or niche could be found. The stapes and the stapedius tendon were present and normal. The ball electrode of the single channel implant was sited in a shallow depression drilled out where the normal anatomical round window should have been. This was held in position with a small amount of ionomeric bone cement. The reference electrode was buried beneath the temporalis muscle and the receiver held in place with prolene ties. Electrode position was confirmed the following morning by post-operative perorbital skull X-ray (Figure 4).

Switch-on

The device was switched on a month later and with further tuning comfort levels ranged from −10 to −7.1 (dBV) with sensitivity at 2.5 and volume at 3. There were occasional non-auditory sensations during tuning sessions in the form of a discomfort in the throat, neck and ear. This was probably due to the leakage of electrical current into the glossopharyngeal nerve via the tympanic plexus on the promontory where the electrode was sited. This did not diminish his enthusiasm. At subsequent tuning sessions the low frequency levels were reduced to minimize non-auditory sensations.

Post-operative results

There have been several problems with the reliability of the equipment which is not robust enough to cope with the lifestyle of a 10-year-old boy. However, the support from the manufacturers with multiple spare parts has been good. The relevant changes in audiological scores were as seen in Table II. He is a skilled lip reader and is beginning to gain additional information from the implant as is borne out by the scores.

TABLE II

Functional listening	Pre-implant	Post-implant
• Syllable pattern (closed set of 12 words)		
Lip pattern and voice:	100%	80%
Voice alone:	0%	42%
• Manchester Picture Test (voice alone)	0%	30%

Discussion

Congenital sensorineural deafness

Jackler *et al.* have proposed a classification of congenital malformations of the middle ear based on embryogenesis (Jackler *et al.*, 1987) in order to differentiate 'true Mondini' deformities from the other varieties. The Mondidi type inner ear dysplasia is probably the commonest variety of cochlear dysmorphology causing sensorineural deafness seen in the clinical setting. Scheibe type malformations are less common. What makes this case of interest is its association with the rare condition of benign familial macrocephaly and infantile hypotonia. It is not clear if the deafness is a separate problem or an unusual manifestation of benign familial macrocephaly. The criteria for diagnosis of autosomal dominant hereditary hearing loss are (1) male to male inheritance pattern; (2) characteristic hearing loss demonstrated audiometrically over three successive generations; and (3) exclusion of other causes of deafness (Kunst *et al.*, 1998). None of these features were seen in this patient and so it can be assumed that this was a chance association and the risk of recurrence of deafness is low but indeterminate.

Benign familial macrocephaly

Benign familial macrocephaly is a rare autosomal dominant disorder with incomplete penetrance (Asch *et al.*, 1976). The features are the same as seen in this patient and CT scans show ventricular dilation in most cases (Cole and Hughes, 1991). The risk of recurrence is one in two.

Related syndromes

Further literature search shows coexistence of macrocephaly and hypotonia has been noted in benign familial macrocephaly, Ruvaclaba-Myhre-Smith syndrome and Bannayan-Zonana syndrome (DiLiberti, 1992). It has been postulated that the above three disorders may be represented in the same gene locus.

The ptosis appears to have been idiopathic and probably an incidental association.

Vibrotactile device

Varying degrees of inner ear aplasias have been described. There are several instances of leakage of cerebrospinal fluid and meningitis either spontaneously (Parks *et al.*, 1982; Phelps *et al.*, 1993) or after cochlear implantation (Page and Eby, 1997). In our patient this risk was compounded by the fact that the child was suffering from recurrent middle ear infections. So at the initial assessment a vibrotactile device (TRILL) was tried out. Although this gives prosodic information about the rhythm and length of sounds, the equipment is cumbersome (especially for children) and was soon discarded.

Multichannel cochlear implant and risks

Multichannel cochlear implants have been inserted in cases with mild bilateral Mondini deformities (Silverstein *et al.*, 1988). But in severe cochleosaccular aplasia there is a definite risk of development of spontaneous cerebrospinal leak or recurrent attacks of meningitis because of wide direct communication between the middle ear and the subarachnoid space in the IAM. Therefore an extracochlear single channel device with an ear level speech processor was considered the only feasible option.

MRI scan

It was also necessary to assess the anatomical integrity of the auditory pathway medial to the aplastic cochlea. It has been seen at autopsy that an intact eighth nerve may be present despite a negligible end organ in a single tube cochlea (Phelps, 1992). With the newer MRI techniques it was possible to demonstrate this in our patient.

Middle ear abnormalities

Anomalies of the round window are also common in subjects with dysplastic inner ears as this develops from the otic capsule and not the branchial arches (Okuno and Sando, 1988). This observation was also corroborated by our findings at surgery.

Single channel cochlear implant

Unlike a vibrotactile device a single channel cochlear implant provides information on pitch changes up to 300 Hz in addition to prosodic information and this would allow improved speech discrimination and environmental awareness (Aleksy, 1983). One of the key factors considered before implanting this child was the need for sound awareness as a safety issue in a farm environment. Not only can he now hear a tractor engine but can reliably turn his head when his name is called.

Current situation

His spontaneous speech contains some vowel sounds and he is beginning to use t,d,k and g. He now has some awareness of his own voice and is beginning to monitor final consonants. Speech intelligibility has improved since the implant was activated. He is able to make himself partly understood to sympathetic strangers. He has won the annual award at his residential school for the deaf for the most improved speech.

Conclusion

Severe sensorineural hearing loss due to severe cochlear dysmorphology can be ameliorated by single channel extracochlear devices without the risks of cerebrospinal fluid leak or iatrogenic meningitis.

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Address for correspondence:
Mr J. Ray, M.S., F.R.C.S.,
89 Longfleet Road,
Poole BH15 2HP.